Genetics Problems

Single trait crosses

1. **Cystic Fibrosis (CF)** is caused by a mutation in the gene for the protein *cystic fibrosis transmembrane conductance regulator* (CFTR). A deletion of three nucleotides results in a loss of the amino acid phenylalanine. This gene is required to regulate the components of mucus. Although most people without CF have two working copies of the CFTR gene, only one is needed to prevent cystic fibrosis. CF develops when neither gene works normally. Therefore, CF is considered an autosomal recessive disease. If a woman heterozygous for CF has children with a man, and none of their children ever exhibit CF, what is the likely genotype of the father?

2. **Galactosemia** follows an autosomal recessive mode of inheritance that confers a deficiency in an enzyme responsible for adequate galactose degradation. Lactose in food (such as dairy products) is broken down by the enzyme lactase into glucose and galactose. In individuals with galactosemia, the enzymes needed for further metabolism of galactose are severely diminished or missing entirely, leading to toxic levels of galactose-1-phosphate in various tissues, as in the case of classic galactosemia, resulting in hepatomegaly (an enlarged liver), cirrhosis, renal failure, cataracts, brain damage, and ovarian failure. If a man with galactosemia has children with a woman without galactosemia, and 1/2 of their children have galactosemia, what is the likely genotype of the mother?

3. **Sickle Cell Disease** is an autosomal recessive genetic disorder characterized by red blood cells that assume an abnormal, rigid, sickle shape. Sickling decreases the cells' flexibility and results in a risk of various blood complications. The sickling occurs because of a single base substitution mutation in the hemoglobin gene. Sickle-cell disease occurs more commonly in people (or their descendants) from parts of tropical and sub-tropical regions where malaria is or was common. One-third of all indigenous inhabitants of Sub-Saharan Africa carry the gene. Those with only one of the two alleles of the sickle-cell disease are more resistant to malaria, since the infestation of the malaria parasite is halted by the sickling of the cells which it infests. Therefore, they may show some symptoms of sickle cell disease but have the benefit of being resistant to malaria. If a man with sickle cell disease (but not malaria) has children with a woman who has malaria, what would be the likely proportions of genotypes and phenotypes of their children?

4. **Phenylketonuria** (PKU) is an autosomal recessive metabolic genetic disorder characterized by a deficiency in the hepatic enzyme phenylalanine hydroxylase (PAH). This enzyme is necessary to metabolize the amino acid phenylalanine to the amino acid tyrosine. If the condition is left untreated, it can cause problems with brain development, leading to progressive mental retardation, brain damage, and seizures. A woman that had PKU as a child has children with a

man who has not ever shown symptoms of PKU. If they have 4 children and 2 are normal, while 2 exhibit signs of brain damage, what are the likely genotypes of both parents?

5. **Huntington's disease** (HD) is a progressive neurodegenerative genetic disorder that affects muscle coordination and also leads to cognitive decline and dementia. It typically becomes noticeable in middle age. The disease is caused by an autosomal dominant mutation on either of an individual's two copies of a gene called Huntingtin. The gene has been recently discovered to be located on chromosome 4 and can now be tested for. The Huntingtin gene normally provides the genetic information for a protein that is also called "Huntingtin". The mutation of the Huntingtin gene codes for a different form of the protein, whose presence results in gradual damage to specific areas of the brain. If a man who later develops HD has children with a woman without HD, and half of their children develop HD as adults, what are the likely genotypes of the parents?

Special Scenario Problems

6. In a certain species of cactus, the plants can have single spines, 2-pronged spines, or a mixture of single and 2-pronged. If 2 cacti showing a mixture of spines are crossed, predict the likely genotypes, phenotypes and corresponding proportions. What type of special inheritance pattern is this?

7. A blue flowered plant is crossed with a yellow flowered plant. All of their offspring have green flowers. If 2 of these green flowered plants are crossed, predict the likely genotypes, phenotypes and corresponding proportions of the offspring. What type of special inheritance pattern is this?

8. Fur color in some cats is determined by a gene located on the X-chromosome. These cats can have yellow patches, black patches, or both black and yellow patches, referred to as a "Calico" cat. Make a punnett square and explain why there could never be a male calico cat.

9. Hemophilia is a disorder that impairs the body's ability to control blood clotting which is used to stop bleeding when a blood vessel is broken. Hemophilia is a sex-linked disorder on the X chromosome. The occurrence of this disorder is about 1 in 8,000 births, mostly males. Hemophilia lowers blood plasma clotting factor levels of the coagulation factors needed for a normal clotting process. Thus when a blood vessel is injured, a temporary scab does form, but the missing coagulation factors prevent fibrin formation, which is necessary to maintain the blood clot. A hemophiliac does not bleed more intensely than a normal person, but can bleed for a much longer time. A male and female who do not display hemophilia have 4 children, 2 boys and 2 girls. Out of the children, only one boy exhibits hemophilia. Explain these results by describing the inheritance pattern and genotypes of the parents and all the offspring.

10. 5 patients are in the hospital and in need of blood transfusions. Patient 1 has type AA, Patient 2 AO, patient 3 OO, patient 4 BB and patient 5 BO. Of the available donors, donor 1 is type AA, donor 2 is type AB and donor 3 is type BO. Use this data to explain which patients can receive blood from which donors…and if any patients are out of luck! Also, state the inheritance pattern of the blood groups.

Two-trait crosses

11. In guinea pigs, short hair is dominant to long hair. Also, black eyes are dominant to red eyes. If a male heterozygous for both traits, is crossed with a female with long hair and red eyes, what are the expected genotypes and phenotypes and in what proportions?

12. Using the same information from above, predict the expected genotypes and phenotypes of a cross between a male guinea pig homozygous recessive for both traits and a female heterozygous for both traits.

Chi Square Problem

13. Albinism is a simple recessive trait in mice, resulting in red eyes versus the usual black eyes. 2 heterozygous mice are mated and produce 72 black eyed offspring and 12 albino offspring.

phenotype	# observed (o)	# expected (e)	(o – e)	$(o - e)^2$	$(o - e)^2 / e$
black					
albino					

$X^2 = \sum [(o - e)^2 / e] =$ _____

Critical values of the Chi square distribution

Probability (p)	Degrees of Freedom (df)				
	1	2	3	4	5
.05	3.84	5.99	7.82	9.49	11.1

df = # of phenotypes – 1

Critical value = _____

Determine whether or not these data are statistically significant enough to support this as a good example of a simple recessive inheritance pattern.

_____
_____
_____
_____

Linked Genes Problem

14. In fruit flies, the genes for singed bristles and blue body are known to be located on the same chromosome; however, the type of inheritance pattern is unknown.

- A wild type female was crossed with a singed bristle/blue male and produced the following F1 offspring:

621 wild-type females	635 wild-type males

- An **F1 female** was mated with a **Parental-male** and produced the following F2 offspring:

355 wild-type males	287 wild-type females
14 singed males	16 singed females
13 blue males	19 blue females
226 blue/singed males	283 blue/singed females

What is the distance between the genes for body color and bristle type?

Calculate the chi square for the null hypothesis that these 2 genes assort independently (There is NO difference in inheritance pattern)

Decide whether to accept or reject the null hypothesis and support with your Chi Square.